



COLEC11 gene

collectin subfamily member 11

Normal Function

The *COLEC11* gene provides instructions for making a protein called CL-K1. This protein is involved in a series of reactions called the lectin complement pathway, which is thought to help direct the movement (migration) of cells during early development before birth to form the organs and systems of the body. The lectin complement pathway appears to be particularly important in directing the migration of neural crest cells, which give rise to various tissues including many tissues in the face and skull, glands that produce hormones (endocrine glands), and portions of the nervous system. After birth, the lectin complement pathway is involved in the immune system.

Health Conditions Related to Genetic Changes

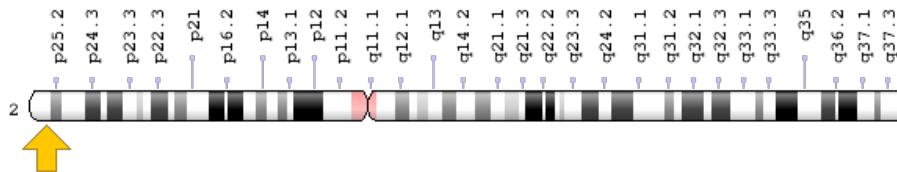
3MC syndrome

At least five *COLEC11* gene mutations have been identified in people with 3MC syndrome, a disorder characterized by unusual facial features and a variety of problems affecting other tissues and organs of the body. The *COLEC11* gene mutations that cause 3MC syndrome impair or eliminate the function of the CL-K1 protein, resulting in faulty control of cell migration in embryonic development and leading to the various abnormalities that occur in this disorder. Researchers suggest that the existence of parallel pathways in the immune system that can compensate for problems in the lectin complement pathway account for the absence of immune system problems in 3MC syndrome.

Chromosomal Location

Cytogenetic Location: 2p25.3, which is the short (p) arm of chromosome 2 at position 25.3

Molecular Location: base pairs 3,594,832 to 3,644,644 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 3MC2
- CL-K1
- CL-K1-I
- CL-K1-II
- CL-K1-IIa
- CL-K1-IIb
- CLK1
- collectin K1
- collectin kidney protein 1
- MGC3279

Additional Information & Resources

Educational Resources

- Immunobiology (fifth edition, 2001): The mannan-binding lectin pathway is homologous to the classical pathway
<https://www.ncbi.nlm.nih.gov/books/NBK27100/#A170>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28COLEC11%5BTIAB%5D%29+OR+%28collectin+sub-family+member+11%5BTIAB%5D%29%29+OR+%28%283MC%5BTIAB%5D%29+OR+%28CL-K1%5BTIAB%5D%29+OR+%28CLK1%5BTIAB%5D%29+OR+%28Collectin+K1%5BTIAB%5D%29+OR+%28collectin+kidney+protein+1%5BTIAB%5D%29+OR+%28collectin-11+isoform+a+precursor%5BTIAB%5D%29+OR+%28collectin-11+isoform+b%5BTIAB%5D%29+OR+%28collectin-11+isoform+c+precursor%5BTIAB%5D%29+OR+%28collectin-11+isoform+d+precursor%5BTIAB%5D%29+OR+%28collectin-11+isoform+e+precursor%5BTIAB%5D%29+OR+%28collectin-11+isoform+f+precursor%5BTIAB%5D%29+OR+%28collectin-11+isoform+g%5BTIAB%5D%29+OR+%28collectin-11+isoform+i%5BTIAB%5D%29+OR+%28collectin-11+isoform+j%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D>

OMIM

- COLLECTIN 11
<http://omim.org/entry/612502>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=COLEC11%5Bgene%5D>
- HGNC Gene Family: C-type lectin domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/1298>
- HGNC Gene Family: Collectins
<http://www.genenames.org/cgi-bin/genefamilies/set/491>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=17213
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/78989>
- UniProt
<http://www.uniprot.org/uniprot/Q9BWP8>

Sources for This Summary

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Reviewed: December 2015

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services